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A CASE OF HEREDITARY MUSCULAR DYSTROPHY.[1]

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THE patient whose case is to be reported is a woman forty-five years of age, in comfortable circumstances. She has always been in good general health, and has not been exposed to severe muscular fatigue. In brief, her early history offers no explanation of her present symptoms. She can, however, remember that even as a child she was unable to run and jump like other children.

When she was about seventeen years old the failure of muscular power became prominently noticeable, and it has slowly but steadily increased ever since.

Her condition at present is as follows :

She walks excessively slowly, with a waddling gait, using her legs as little more than crutches. When she throws her whole weight upon either foot, as in going up or down stairs, the foot turns so that the leg almost rests upon the ground, and she is wholly unable to go up or down stairs without either clinging with her hands to the balustrade, or going on all fours.

The hands and arms are still useful members, though feeble. When examined more closely, it is found that the movements of the feet and toes are practically though not absolutely abolished, but that all others are possible to a greater or less degree.

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[1] This case was reported in connection with the paper upon "Muscular Dystrophies," read by Dr. Sachs at the last meeting of the Neurological Association.

The thigh-trunk muscles are excessively feeble, as is shown by the manner in which the trunk is thrown from side to side when the patient walks. She can, however, sit erect, and stands without marked lordosis.

Her best movements are those of the shoulder, and the muscles there appear to be in the best condition. The muscles below the knees are excessively wasted, but rather dense, and with the skin drawn tightly over them.

The biceps of both sides are also much wasted, looking as if hollowed out at its middle portion.

The bellies of the flexor muscles of both forearms are short and rather prominent, and slightly dense to the touch, while, at the same time, they are deficient in strength, as is shown by the feebleness of the patient's grasp, and by the fact that the effort is attended by a throwing back of the hand into position of hyperextension.

There is no sign of muscular hypertrophy, except as indicated by this relatively dense consistency of the flexors of the arms, and the fact that these muscles, although short and feeble, are more rounded and prominent than would be expected.

It will be shown later that apparently hypertrophied fibres were, however, met with not only in these muscles but also in others where no outward trace of any hypertrophic process was to be seen.

The patient's skin is of a dark color, and has been for more than twenty years the seat of chronic psoriasis. The dark color is not, however, confined to the diseased portions.

The face is of the same dark tint with the rest of the body, and the muscles there are also rather feeble, although all motions are possible. In masticating food the muscles of the jaw feel weak and get fatigued. The intrinsic muscles of the hand are well preserved.

The patient, who is a very intelligent person, complains that for a long time past she has had indescribable sensations in her head, which, she says, makes it impossible for her to collect her thoughts. She also falls down occasionally, not from tripping or losing her balance, but in consequence of some cerebral cause, although she feels sure that she does not wholly lose consciousness. Ever since seven or eight years old she has found increasing difficulty in committing to memory.

The electrical examination, which was made with great care, gives the following result:—

Faradic current; facial nerves; separation of coils necessary for mimical response, 120 mm. (that is, a very weak current, the reaction being normal); ulnar nerve, separation of coils, etc., 123 mm.; media nerve, separation of coils, 105 mm.

The muscles of the thenar eminence react at 120 mm.; Exten. dig. long. at 105 mm.; flexors of forearm at 105 mm.; biceps at 105 mm. (contractions very feeble).

In other, words the facial nerve, the small muscles of the thumb, and the ulnar nerve show a normal reaction. The reaction of the median nerve is apparently diminished, but probably on account of the condition of the flexor muscles. The reactions of the biceps and of the flexors of the forearm are also diminished.

The muscles of the lower extremity below the knee respond only to maximal currents, upwards of 70 mm., when the electrode is applied to the nerve, while a still stronger current than this is required to make the muscles contract by local stimulation. The reaction of the ex. quad. long. is slightly better, but the contraction is excessively slow and feeble. The reactions to the galvanic current are exactly proportionable to those of the faradic. No trace of degenerative reaction is anywhere seen.

The patient reports, with regard to her *family history*, that her father was a vigorous man, and died of cancer at the age of seventy.

Her mother was in good health, but had three paralytic attacks in late life, and died at eighty-six.

The father had two brothers. One of them began at seventeen to have similar troubles with that of the patient, and after a time became entirely crippled, so that he could not even hold his head erect, or use any part of the body, but he lived for several years in this condition, and lived to be over sixty years old. The other brother was well until about twenty-five years old. Then he had some weakness which was called rickets, and from this he never recovered. He became gradually helpless, so that he could not stand or walk, but could wheel himself around in a chair. He lived to be about sixty, and died within a day or two of the elder brother above mentioned.

The patient's paternal grandfather died from injury at sixty-five years. Health good.

The paternal grandmother died from a paralytic shock at seventy. This grandmother had a sister who became a cripple. The patient does not know at just what age her weakness came on, but she was finally so helpless that she had to be lifted in and out of bed. The family history back of this point is not known.

The patient has a brother who was in the army, and, of late, has lost his health, and is supposed to be consumptive. She has recently heard from him that he is losing the power in his legs, but cannot say exactly to what extent.

The patient had one sister who died of consumption. Four brothers have also died, but mainly from acute affections. On the mother's side the family were not considered very healthy. One branch was liable to what was called "numb palsy."

The psoriasis from which the patient herself suffered is said to have occurred in the mother's side of the house.

For the five months previous to June, 1888, the patient was treated daily with strong applications of electricity, the combined F. and G. current being used, and has also taken phosphorus, cod liver oil, and arsenic most of the time. Under this treatment there was a marked improvement, which seems to be well established.

The patient can walk very much longer distances than before, and with less fatigue. She can get her hands behind her back so as to apply ointments, etc., which was at first almost impossible, and can use her hands for sewing and knitting, and feeding herself, all of which were difficult and fatiguing before the treatment was begun. Moreover, the universal psoriasis had, by June, practically disappeared, there being only a few spots on the back and on the feet remaining.

This disappearance has been gradual, having begun about February or March. The abscesses also ceased reforming early in the spring. The feeling of confusion and distress in the head also became less, especially during the last few weeks of treatment, when galvanism was applied to the head.

During the summer she was in the country, and soon discontinued the arsenic and electricity, but kept on with the phosphorous and oil.

The gain in strength maintained itself, but the distress in the head increased, and the psoriasis returned. I attribute the previous gain mainly to improvement in nutrition and nervous energy, through the tonic, and, in part, through the electrical treatment, rather than to a definite change in the muscular condition.

The electrical investigation led me, however, to think highly of the use of the combined faradic and galvanic currents, as recommended by DeWatteville, which made it possible to obtain contractions with weaker faradic currents than would otherwise have been necessary.

It was noticeable that the contractions obtained in this way were increased, not only at the moment of closure of the galvanic circuit, but during the flow of the current.

As regards the psoriasis, its disappearance may of course have been accidental, but as this had never happened before, within the patient's memory, it seems fair to attribute it to the arsenical treatment.

Two pieces of muscle were removed for examination; the first from the right biceps, at the part where the muscle was so much atrophied that a sort of excavation had taken the place of the usual bulging of the muscle; the second from the right flexor com. dig.

The biceps was one of the muscles that still responded to voluntary effort, and also to the faradic current, but slowly and feebly. The reaction to the galvanic current was also diminished, but there was no excessive action of the positive pole, nor was the contraction slower than under the faradic current.

The forearm muscle contracted better than the biceps, where the cut was made, but still slowly and feebly. The belly of the muscle was also markedly shorter than normal.

The biceps muscle was examined fresh, and also after hardening in Müller's fluid. The fresh specimens showed here and there a partial or complete loss of transverse striation, though most of the fibres had preserved their striations.

Many of the fibres were marked longitudinally by fine granules arranged in lines and spindle-shaped forms, and consisting of fat probably.

Another change also seen in both fresh and hardened specimens was a tendency to split up into fibrillæ, some of which showed transverse striations, and some no striations.

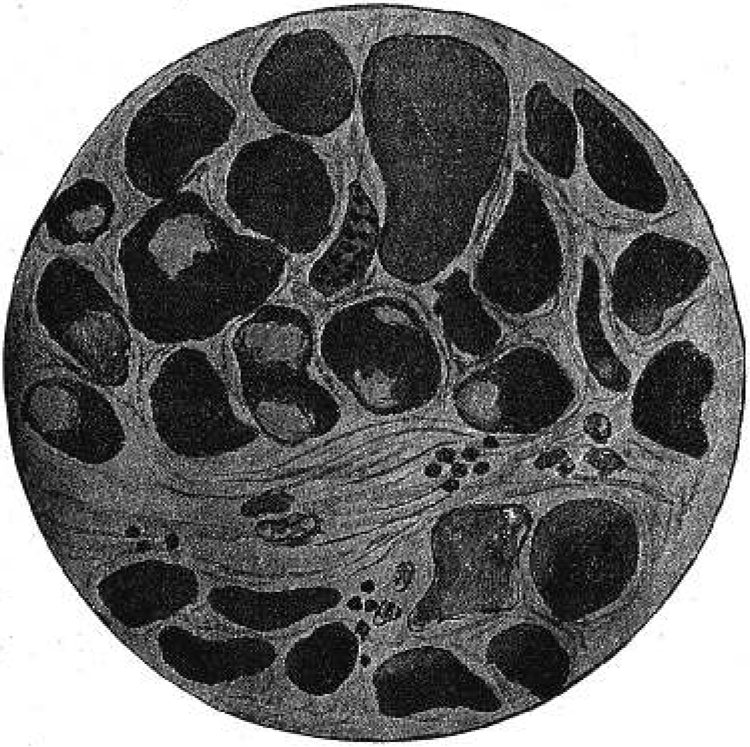
The hardened specimen was cut in both longitudinal and transverse directions, and sections stained in various ways.

The transverse sections showed, first, a very striking variation in size between individual fibres, the smallest being less than half the size of ordinary muscle fibres; the largest at least twice as large as normal fibres. The corners and edges of these fibres instead of fitting together, with fine lines between them, are rounded off and more or less widely separated, the interstices being filled up by connective tissue, containing here and there a large number of cells, and in other places but few cells.

The shape of the individual fibres also varies greatly, not only because of the direction in which they happen to be cut, but also because a portion of the fibre is often destroyed.

The muscular bundles are also widely separated by connective tissue, which often contains glistening yellowish masses, varying in size from that of a pus cell to that of a muscular fibre. When examined separately a large proportion of the muscular fibres appear to be altered.

They are marked longitudinally by fine lines, as if teased with a comb, and the fibre is thus broken up into smaller or larger fibrillæ.



There is also evidence of a gross destructive process which has sometimes gone so far that the fibre appears as if the central portion had been destroyed, leaving a cavity behind.

In other places the fibre looks as if only irregularly thinned toward its centre.

Many of the fibres have a practically normal appearance. Here and there a place is seen even in the midst of a muscular bundle, where a fibre seems to have been entirely destroyed, only its outline being left, filled with cells which are about the size of muscle cells, though varying somewhat in dimensions. The blood vessels appear, on cross sections, to be surrounded with round cells to a moderate degree.

On longitudinal sections the most striking appearance is that of the splitting up of the muscle into fibrillæ, both where the section cuts obliquely across the end of a muscular fibre, where the fibrillæ appear frayed out, as by a comb, and also in the course of the fibre, giving rise to a marked longitudinal striation, with sometimes an appearance as if the fibre had been scored with a knife.

Most of the fibres, even where this change has taken place, have preserved their transverse striation.

There is almost everywhere a great increase of the interstitial nuclei, but in some places this is greater than in others.

The sarcolemma nuclei are also here and there greatly increased in number, but not uniformly. No intra muscular nerves could be found on these sections.

At rare intervals a fibre was seen which was excessively atrophied, and presented a homogeneous, glassy appearance.

An appearance is here and there met with as of a protoplasmic mass containing several small nuclei. Apparently this is in place of a muscular fibre which has been destroyed and represents its residue.

The piece taken from the forearm was examined fresh, and also after treatment with chloride of gold and formic acid, this method being chosen in the hope of finding the terminal muscular plates.

It may be said, in brief, that a similar difference in the size and shape between the muscular fibres was found as in the fresh specimens. On the whole, however, the alterations were much less strongly marked.

A great majority of the muscular fibres had preserved their transverse striation.

It was noticeable that the hypertrophic fibres were by no means always those in which the destructive changes were most strongly marked. A small nerve fibre was found in one preparation, and traced up to the muscle, but no terminal plates were seen. The nerve fibre itself seen appeared to be perfectly healthy.